



HYPERMETHIONINEMIA (MET)

FAMILY FACT SHEET

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 36 hours after birth. The blood is tested for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby might have a disorder called hypermethioninemia.

A positive newborn screen does not mean your baby has hypermethioninemia, but it does mean your baby needs more testing to know for sure.

You will be notified by your primary care provider or the newborn screening program to arrange for additional testing.

What is Hypermethioninemia?

Hypermethioninemia affects an enzyme needed to break down proteins in the food we eat, so they can be used for energy and growth. In hypermethioninemia, the enzyme used to break down proteins is missing or not working properly.

A person with hypermethioninemia doesn't have enough enzyme to break down protein containing methionine. When the body can't break down methionine, it builds up in the body and causes health problems.

Hypermethioninemia is a genetic disorder that is passed on (inherited) from parents to a child. The mother and father of an affected child carry a gene change that can cause hypermethioninemia. Parents usually do not have signs or symptoms, or even know they carry the gene change.

What problems can Hypermethioninemia cause?

Hypermethioninemia is different for each child. Some children with hypermethioninemia have few health problems, while other children may have a more severe form of hypermethioninemia with serious complications.

If hypermethioninemia is not treated, a child might develop:

- Sleepiness
- Weak muscle tone
- Developmental delay
- Liver problems

It is very important to follow the doctor's instructions for testing and treatment.

What is the treatment for Hypermethioninemia?

Hypermethioninemia can be treated. Treatment is life-long and can include:

- Low protein diet - a dietician will help you set up the best diet for your child.
- Medications (Vitamin B6) to help break down proteins.

Children with hypermethioninemia should see their regular doctor, a doctor who specializes in hypermethioninemia, and a dietician.

Prompt and careful treatment helps children with hypermethioninemia live the healthiest lives possible.

Michigan Resources and Support

Michigan Newborn Screening

Nurse Consultant

Toll-free: 1-866-673-9939

newbornscreening@michigan.gov

Children's Hospital of Michigan Metabolic Clinic

Toll-free: 1-866-442-4662

Children's Special Health Care Services

Toll-free: 1-800-359-3722



Michigan Newborn
Screening

Phone 1-866-673-9939

www.michigan.gov/newbornscreening

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